Washington State Department of Health

NEWBORN SCREENING PROGRAM
OVERVIEW
DECEMBER 15, 2004

It is the stated policy of the state of Washington "...*to make every effort to detect as early as feasible and to prevent where possible phenylketonuria and other preventable heritable disorders leading to developmental disabilities or physical defects*" (Revised Code of Washington, RCW 70.83.010; Declaration of policy and purpose). The law gives authority to the State Board of Health to determine which disorders are to be detected and directs the board to adopt regulations as needed to carry out the intent of the law.

Chapter 246-650 WAC requires screening for nine preventable disorders. Universal screening of infants for Phenylketonuria (PKU) was first mandated in 1966. Congenital hypothyroidism (dysfunction of the thyroid gland) was added in 1977. Congenital adrenal hyperplasia, a disease that affects steroid hormone production, was added in 1987 and, in 1991, the Board added sickle cell disease and other hemoglobin disorders such as thalassemias to the list of disorders. Finally, in October 2003 the Board added five disorders of metabolism to the screening regimen: biotinidase deficiency, galactosemia, homocystinuria, maple syrup urine disease (MSUD), and medium chain acyl co-A dehydrogenase (MCAD) deficiency. Testing for biotinidase deficiency and galactosemia was implemented on January 1, 2004 and tests for the remaining disorders began on June 1, 2004.

Board of Health regulations (chapter 246-650 WAC) direct hospitals throughout the state to collect a small specimen of blood from each infant's heel before they are discharged. The specimens are absorbed on a special type of paper, dried, and then sent to the Department of Health's Public Health Laboratories where highly sensitive and specific tests are performed for each of the disorders. The laboratory employs a range of technologies including immunoassay, high performance liquid chromatography, iso-electric focusing, mass spectrometry, and DNA/polymerase chain reaction.

Positive test results are immediately communicated to the child's health care provider along with consultation to link them with appropriate diagnostic and treatment services. The program tracks all positive results to assure that the children receive the necessary care as quickly as possible. The program also tracks the long-term outcome of infants found to be affected by the disorders to evaluate the effectiveness of the system

Over two million infants have been screened since 1966 and hundreds of children have been given the opportunity to live healthy, productive lives because their conditions were detected and treated in time through this screening. These children have been spared permanent disability, mental retardation, or tragic death that would otherwise result from their disorders.

The screening program is self-sustaining through a charge that is collected through the hospital where the child is born. Diagnostic and treatment care is funded through many sources including government and private insurance, federal grants and self-pay. A small fee that is collected at the same time as the screening charge helps subsidize specialty care clinics for the disorders.

For more information call:
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